

Bronwyn Kerr

List of Publications by Year in descending order

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Version: 2024-02-01

24
papers

1,525
citations

361045

20
h-index

610482

24
g-index

26
all docs

26
docs citations

26
times ranked

3844
citing authors

#	ARTICLE	IF	CITATIONS
1	High penetrance of myeloid neoplasia with diverse clinical and cytogenetic features in three siblings with a familial GATA2 deficiency. <i>Cancer Genetics</i> , 2021, 256-257, 77-80.	0.2	0
2	Ligase IV syndrome can present with microcephaly and radial ray anomalies similar to Fanconi anaemia plus fatal kidney malformations. <i>European Journal of Medical Genetics</i> , 2020, 63, 103974.	0.7	5
3	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 103.	1.2	23
4	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. <i>American Journal of Human Genetics</i> , 2020, 106, 596-610.	2.6	59
5	Costello syndrome: Clinical phenotype, genotype, and management guidelines. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1725-1744.	0.7	70
6	Pathogenicity and selective constraint on variation near splice sites. <i>Genome Research</i> , 2019, 29, 159-170.	2.4	70
7	Catalogue of inherited disorders found among the Irish Traveller population. <i>Journal of Medical Genetics</i> , 2018, 55, 233-239.	1.5	19
8	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	2.6	59
9	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	2.6	87
10	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. <i>American Journal of Human Genetics</i> , 2018, 103, 752-768.	2.6	40
11	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 1195-1203.	2.6	37
12	Autism spectrum disorder and other neurobehavioural comorbidities in rare disorders of the Ras/MAPK pathway. <i>Developmental Medicine and Child Neurology</i> , 2017, 59, 544-549.	1.1	40
13	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	2.6	337
14	Further Clinical Delineation of the MEF2C Haploinsufficiency Syndrome: Report on New Cases and Literature Review of Severe Neurodevelopmental Disorders Presenting with Seizures, Absent Speech, and Involuntary Movements. <i>Journal of Pediatric Genetics</i> , 2017, 06, 129-141.	0.3	38
15	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. <i>JAMA Neurology</i> , 2017, 74, 1228.	4.5	79
16	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. <i>American Journal of Human Genetics</i> , 2017, 101, 995-1005.	2.6	23
17	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 1021-1033.	2.6	83
18	Protein structure and phenotypic analysis of pathogenic and population missense variants in STXBP1. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 495-507.	0.6	29

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19	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. <i>American Journal of Human Genetics</i> , 2016, 98, 363-372.	2.6	36
20	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. <i>European Journal of Medical Genetics</i> , 2015, 58, 455-465.	0.7	83
21	Leri's pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassing <i>GDF6</i> and <i>SDC2</i> and provides insight into systemic sclerosis pathogenesis. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 1249-1256.	0.5	22
22	Further delineation of the KAT6B molecular and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2015, 23, 1165-1170.	1.4	56
23	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. <i>Human Molecular Genetics</i> , 2014, 23, 4315-4327.	1.4	114
24	The genetic basis of DOORS syndrome: an exome-sequencing study. <i>Lancet Neurology</i> , The, 2014, 13, 44-58.	4.9	108